A SOCIOBEHAVIOURAL PERSPECTIVE ON GENETIC TESTING AND COUNSELLING FOR HERITABLE BREAST, OVARIAN AND COLON CANCER

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Abstract • Résumé

Testing for susceptibility to heritable breast, ovarian and colon cancer has unique psychosocial costs. Negative test results may not be sufficient to relieve anxiety, and positive results can cause sufficient distress to compromise patient compliance with surveillance and risk reduction measures. More needs to be learned about how sociocultural factors affect the understanding of risk, how decisions to undergo testing are made and how information about increased risk affects family dynamics. As the demand for testing and counselling grows, health care providers will be faced with new challenges and dilemmas. A better understanding of genetics by the public is needed to mitigate deterministic attitudes that can lead to the neglect of health promotion. Also of concern are the socioeconomic implications of being identified as having a high risk for heritable cancer and the dangers inherent in using genetics to explain sociological phenomena. Health care providers must take the lead in ensuring that developments in genetics are used to the benefit of all.

Les tests pour déterminer la vulnérabilité au cancer héréditaire du sein, de l'ovaire et du côlon entraînent des coûts psychosociaux uniques. Il se peut que des résultats négatifs ne suffisent pas à dissiper l'anxiété et que des résultats positifs provoquent une détresse suffisante pour compromettre l'observation par le patient des mesures de surveillance et de réduction du risque. Il faut en apprendre davantage au sujet de la façon dont les facteurs socioculturels jouent sur la compréhension du risque, de la prise des décisions de subir des tests et de l'impact que l'information sur le risque accru a sur la dynamique familiale. À mesure qu'augmentera la demande de tests et de counselling, les fournisseurs de soins de santé seront confrontés à des défis et à des dilemmes nouveaux. Il faut que la population comprenne mieux la génétique afin d'atténuer les attitudes déterministes qui peuvent faire négliger la promotion de la santé. Ce qui préoccupe aussi, ce sont les répercussions socio-économiques du fait d'être identifié comme un sujet à risque élevé de cancer héréditaire et les dangers inhérents au recours à la génétique pour expliquer des phénomènes sociologiques. Les fournisseurs de soins de santé doivent jouer un rôle de premier plan pour assurer que les progrès de la génétique soient utilisés pour le bien de tous.

The identification of genetic markers for heritable breast, ovarian and colon cancer[1–4] has advanced with exceptional speed, and testing to identify people at increased risk in families with heritable cancer syndromes is becoming more readily available. Because the duty of all health care professionals is to provide benefit and minimize harm, they have a responsibility to examine carefully the effect that new capabilities in genetic testing will have on society. We reviewed the relevant social science literature on genetics to determine the impact that information on risk for heritable breast, ovarian and colon cancer will have on health care consumers and providers and the general public. Our analysis of the literature revealed that the sociobehavioural implications of genetic testing for susceptibility to disease are not fully understood; this finding points the way for further research.

SOURCES AND METHODS

We did an extensive, iterative online search of MEDLINE and CANCERLIT and of sociology, psychology,

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humanities and social-sciences databases using an extensive range of key words and phrases. We assessed 1056 articles and included 294 of these in the final database. We categorized and summarized these articles using content analysis techniques.[5] We found few reports of empirical studies that specifically considered the sociobehavioural implications of providing information about risk for heritable breast, ovarian and colon cancer. However, we did find useful information about the psychosocial effects of screening and surveillance programs for breast, cervical and colon cancer. In addition, we were able to extrapolate from the literature on genetic risk for heritable monogenic diseases such as Huntington's disease and Tay–Sachs disease. Given that sociobehavioural research in this area is in its infancy, we made no attempt to appraise the articles critically, and we included reports of both quantitative and qualitative research in the final database.

Overview

Medical practitioners have been able to identify people with certain inherited conditions since the late 1970s. New techniques now permit the identification of people who are susceptible to various types of cancer before disease develops. The application of genetic testing — which previously was limited to the diagnosis of well-defined mendelian syndromes — to susceptibility to cancer creates new clinical, social and economic dilemmas. It also requires a fundamental change in how information about genetic risk is presented to health care consumers. The knowledge that certain people may be at increased risk for a given disease could create, as Nelkin[6] points out, a new "genetic underclass," the "presymptomatic ill." To put the sociobehavioural implications of the provision of cancer risk information into perspective we require an understanding of three key areas: the perspective of the consumer, the response of health care providers and the expectations of the public.[7,8]

The consumer's perspective

Genetic testing is of benefit to consumers presumed to be at risk only if it yields reassuring information or prompts action that will help to prevent cancer or facilitate its early detection. If information about genetic risk has the potential to cause psychological harm, it is crucial that we clearly identify such hazards and evaluate ways to minimize or eliminate them.[9,10]

Many studies have examined the reasons why people want genetic testing. For example, people in high-risk groups may want to be reassured about their own and their children's health risks.[11,12] In one study of the perceived demand for testing for susceptibility to breast, ovarian and colon cancer, most of the people surveyed anticipated that negative test results would improve their quality of life (83%) and make them feel less anxious (83%), less depressed (68%) and more in control (82%).[13] Given the adverse psychological effects that can arise from either false-positive or false-negative results,[13,14] accuracy is crucial to the ability of any test to provide reassurance. For example, investigations of the psychological impact of cancer screening programs showed that a false-positive mammogram result produced acute psychological distress in some women that was sufficient to undermine daily functioning and that persisted long after they were shown not to have breast cancer.[12,15,16]

Studies involving patients tested for Huntington's disease showed that people can have elevated levels of distress after receiving negative *or* positive test results.[17,18] In one study approximately 10% of participants who received negative results still experienced adverse effects 6 months later.[19] In another study 72% of women at risk for familial breast cancer expected to go on worrying even if they were given negative test results for mutations of the *BRCA1* gene.[13] Studies have also reported negative sequelae, notably "survivor guilt" and shame, among family members who were told that they probably did not have an altered *BRCA1* gene.[20–24]

Carrier status has been associated with problems with self-image and feelings of stigmatization.[25–27] Participants in a pilot study who were found to be genetic carriers for Tay–Sachs disease viewed their future health status more negatively than noncarriers.[28] Judging from these results, it would seem likely that a positive test result for cancer susceptibility would have adverse effects on how people perceive themselves and are perceived by others.

Increased worry about cancer among women at high risk for familial breast cancer was found to interfere with adherence to surveillance measures such as breast self-examination and attendance for clinical breast examination and mammograms.[29] Such findings raise the concern that knowledge of positive results from a genetic test could also adversely affect participation in activities that could potentially reduce risk and prolong life.

People found to be at high risk for a genetic disease may be concerned about the fate of their children and siblings. The effect of testing for cancer susceptibility on family relationships is not yet fully understood. Relatives may have both accurate and inaccurate information about cancer and are likely to differ in their degree of scientific and genetic literacy. They may also be concerned about the social and economic ramifications of the identification of a gene for cancer susceptibility in their family.[30,31]

Sociocultural factors affect how consumers process information about cancer risk. If such information con-

flicts with basic beliefs and cultural values it may be perceived as useless, shameful or as precipitating a negative outcome.[32–35] There is a high degree of family orientation in many cultures, and counselling and decision-making requiring informed consent may need to take place in a family context.[36] As well, certain genetic diseases are more prevalent in particular ethnic or racial groups. In the past, discriminatory practices have been directed toward certain ethnic or religious groups in the guise of genetic screening and counselling for disease (e.g., sickle-cell anemia in black Americans).[37–39] Care is needed to ensure respect for cultural norms and values.

Gaps in our understanding of the psychological impact of testing for susceptibility to disease pose many questions. For example, to what degree will notification of high risk affect a low-risk spouse, and how will the marital relationship be affected? Would parents who are notified of a child's high risk treat that child differently? What effect will there be on the psychosocial development of children who are told at an early age that they are at high risk? What factors motivate or dissuade some people to undertake testing for heritable cancers? What are the psychological effects of recommended prophylactic and chemopreventive interventions?

The response of health care providers

Health care providers have many issues to consider as they deal with the expectations aroused in consumers by the availability of tests for cancer susceptibility. Although testing for cancer susceptibility and for other types of genetic disease create a similar need for counselling, there are key differences in the expression of these diseases and in the potential for their treatment. Moreover, a negative test result for cancer susceptibility does not guarantee that the person tested will never get cancer.

To complicate matters, providers may be approached by third parties such as insurance companies, employers and family members to disclose confidential genetic information. These pressures could adversely affect how much information about risk is given, the decision-making process and the confidentiality of the relationship between patients and health care providers.[40]

The accepted standard in genetic counselling for mendelian disorders and issues related to reproduction, prenatal diagnosis and pregnancy termination has been one of "nondirectiveness" whereby direct advice is withheld by the counsellor. However, questions have been raised as to whether it is possible or appropriate to be truly nondirective.[41–44] The optimal continuum of genetic counselling with respect to risk for breast, ovarian and colon cancer would possibly include not only providing information but also exploring the implications of that information as well as giving specific, directive medical advice.[12]

The use of a more prescriptive approach to genetic counselling for people undergoing genetic testing for breast, ovarian and colon cancer requires that patients are enabled to make informed decisions. It is essential that consumers understand the potential limitations, risks and benefits of genetic tests, and their personal, familial, economic and social consequences. Patients do not always fully comprehend the information provided in consent forms.[45–47] One study conducted in a large genetic counselling centre[48] revealed that of the 87% of clients who had a faulty understanding of genetic risk before counselling, over half still had misconceptions after counselling. Of the 13% who had entered with accurate information, almost one-fifth left with misconceptions. These findings emphasize the importance of presenting information clearly in several different ways and of shifting the focus from consent forms to the development of appropriate consent *processes*.[49–54] It has been recommended that informed consent be obtained through full disclosure of all costs, risks, benefits and limitations of genetic screening within a pretest and post-test counselling protocol.[12,20,21,24]

The provision of comprehensive genetic testing and counselling has major implications for resource allocation. Difficulties related to the delivery of appropriate services were identified in a comprehensive survey involving 18 countries.[55,56] A recent study by the Science Council of Canada showed that over 80% of existing Canadian genetic centres were unable to meet current demands for service and predicted that the problem would worsen.[57] As genetic tests become more readily available, there will be an even greater need for counselling, which is time-consuming and not cost recoverable.[58–60] As more people demand testing, financial and staffing constraints will allow proportionately fewer consumers to receive the counselling they need to understand and cope with their test results.

Currently, clinical and medical geneticists and genetic counsellors provide most genetic counselling services. As more tests become readily available, more personnel will be required. Although primary care physicians, given their ongoing relationship with patients and their families, may be ideally suited to this role, they will require considerable training in genetics and counselling techniques.[57,61–66]

Studies assessing genetics teaching in medical schools and physicians' knowledge of the subject have found that both are inadequate.[57,67–70] Primary care physicians whose knowledge was assessed were able to answer less than 75% of questions relating to the genetic concepts and facts required to offer genetic testing and counselling.[69] Physicians may not have the psychological

training needed to assist patients who have been given positive test results or to address the psychosocial impact on families. Time constraints, financial limitations and concerns about medicolegal liability, and a relatively low tolerance for diagnostic ambiguity, may also impede the ability of primary care physicians to provide optimal genetic counselling.[71-73]

Other kinds of health care providers will be needed to meet the increasing demand for counselling. Registered and public health nurses, social workers and psychologists could, with the appropriate training, fill the gap. To overcome cultural and religious barriers to effective counselling, it will be important to train providers from diverse cultural backgrounds and to integrate cross-cultural education into training programs.[74-76]

Expectations of the public

North Americans are understandably intrigued by media reports of developments in genetics, but their scientific literacy is not improving concomitantly.[57,77] In one study educated laypeople who were tested on their understanding of a selection of media reports on health research had a rate of error of close to 40%.[78] Many people do not understand the principles of probability, which makes it difficult for them to comprehend risk estimates.[79-81] The nature of the public's awareness of genetic disease and genetic testing raises further concerns: many people are more familiar with genetic defects that result in developmental disability (e.g., Down syndrome) than with those that result in physical impairment, and information about genetic testing is not widely disseminated.[82,83]

Public attitudes toward genetic testing for susceptibility to disease have not been widely investigated. Studies of attitudes of people at risk for Huntington disease toward presymptomatic testing may provide some insight. Study participants who chose not to be tested refused because of major concerns related to the lack of a cure, the potential loss of insurance coverage and the effect of positive results on their children.[84] In contrast, participants in a study on attitudes toward testing for susceptibility for colon cancer had a high level of interest, especially when they thought themselves to be at risk.[85] Thus the desire for testing seems to depend on the perception of the potential for a cure.

The overall conclusion is that the general public requires a better understanding of genetics to evaluate scientific assertions about susceptibility to cancer. Health care providers will need to assume the responsibility of identifying the essential components of genetic literacy.[57,61] A broader understanding by society of the potential and the limitations of genetic testing might help to defuse the notion that human destiny is genetically determined. Without this understanding the public may conclude that important efforts such as environmental protection, health promotion and cancer prevention are futile.

Implications for the future

As future directions for research and policy development are considered, important psychological and socioeconomic issues need to be addressed.

Psychological costs, risks and benefits

There are well-established protocols for the increased surveillance of people at high risk for breast, ovarian and colon cancer. These include regular breast self-examination, clinical breast examination, mammography, transvaginal ultrasound screening and colorectal cancer screening. However, whether cancer mortality among high-risk groups is at all reduced by increased surveillance remains unclear.[21,86] Research focusing on strategies that will promote adherence to surveillance measures by people who are at high risk is needed. Such strategies should address known barriers to patient compliance, such as anxiety about radiation and investigative procedures. Interventions should promote a secure, emotionally supportive setting for surveillance in which embarrassment and anxiety are minimized.

Preventive options for people at risk for heritable breast, ovarian and colon cancer include prophylactic mastectomy, oophorectomy, colectomy and chemoprevention. The psychological costs of prophylactic mastectomy in healthy women who believe they are at risk for breast cancer have yet to be assessed but may be mitigated by the active choice involved, the alleviation of anxiety about cancer and a reduced need for surveillance with its attendant costs.[12,21,87,88] Health care providers, however, should be wary of unduly influencing healthy women to undergo such radical procedures, given the lack of convincing evidence of their benefit.

Prophylactic oophorectomy has been recommended for women with a 50% or greater lifetime risk of ovarian cancer. Although the protective effects of oophorectomy have been questioned because of reports of intra-abdominal carcinomatosis occurring after surgery, this risk may have been overestimated. However, several medical risks are associated with early menopause, as well as with long-term hormone replacement. Moreover, oophorectomy has potential psychological costs, especially for women of childbearing age.[89-92] It remains unclear how women at risk for ovarian cancer evaluate the potential personal costs of preventive oophorectomy against the relief of reducing their risk of cancer.

Women at high risk for ovarian cancer are advised to

consider oral contraceptive therapy to decrease their risk.[93,94] In addition, women at high risk can enrol in the Tamoxifen Chemoprevention Trial.[95] However, recent media reports of an association between increased risk of uterine cancer and tamoxifen use have raised additional concerns among women already worried about cancer. Although there may be a degree of psychological morbidity among women receiving tamoxifen therapy, no studies have specifically addressed the psychological costs or benefits of chemoprevention.

Colectomy is recommended for teenagers from families with familial adenomatous polyposis (FAP) syndrome. Those who test positive for this disorder have a nearly 100% risk of developing colon cancer unless the colon is surgically removed. There has been only limited study of the long-term psychological effect on young adults of such interventions.[96–98]

The general maxim in health care is "the earlier the diagnosis the better," but testing to assess susceptibility to disorders for which there is no treatment until the onset of disease (and perhaps not even then) may be of no benefit to young people.[99–102] Indeed, there may be damaging psychosocial repercussions. In the extreme, public knowledge of a person's test results may render him or her "uninsurable, unemployable and unmarriageable."[61]

Socioeconomic issues

Genetic testing creates new categories of people: those who have genetic "abnormalities" and those who do not. It is easy to understand how criteria of efficiency and cost containment could be used to segregate society into "good-risk" versus "bad-risk" groups. Although some people argue that sufficient protection exists to prevent genetic discrimination, others maintain that current legislation may not cover new circumstances that will arise out of genetic testing.[57,61,103,104]

Private-sector organizations such as insurance companies and employers, and public-sector institutions such as schools, health care organizations and government are seeking ways to lower costs and maximize resources and will take great interest in genetic testing. Genetic testing introduces a fundamentally new way of assessing risk and may be perceived by underwriters as another tool to predict insurers' financial risk.[105] Job discrimination resulting from genetic testing is also a growing concern. Employers trying to control the costs of employee benefit packages for life, disability and health care plans may choose not to employ people whom they think have increased health risks.[106–108]

Genetic predisposition has been suggested as the cause of a wide range of problems, including homosexuality, alcoholism, smoking behaviours, schizophrenia and learning problems.[38,109–112] This trend toward genetic determinism — the transformation of social problems into essentially medical problems — opens the door to a new form of social eugenics, creates new social classes based on genetic inheritance and may divert attention and funding from the systemic and environmental causes of social problems.[6,7,38,113–117]

People are now being asked to make choices, based on genetic tests, that will affect their own and their children's health and, by implication, society's health care costs. People still have the right to decide whether to undergo genetic testing, but if genetic testing or screening becomes public policy, people could lose the right to make their own decisions.[118–122]

Despite the harm that could be associated with genetic testing, the demand for population-based screening for susceptibility to breast, ovarian and colon cancer could be substantial, given the estimated carrier rate for *BRCA1* mutations (1 in 400 to 1 in 200 women).[2,3,123,124] The resources available for genetic counselling and testing will limit the number of tests that can be performed. Health care professionals and policymakers will need to decide whether general screening for cancer susceptibility should be promoted through the use of public policy interventions, or whether selective screening (with clearly defined inclusion criteria) should be offered.

Conclusion

Promising new developments in cancer genetics are harbingers of future discoveries that have implications beyond their clinical utility. As our capabilities in genetic testing increase, there appears to be a growing societal trend toward "genetic reductionism" and an inherent danger of providing simplistic explanations for multifaceted problems and thus letting society "off the hook." If, for example, businesses can blame carcinogenesis on genetic factors, they may not be inclined to accept responsibility for monitoring and reducing environmental contamination. If teachers can use genetic rationalizations to account for poor learning performance, they need not question the effectiveness of current educational practices. Society is absolved of responsibility for improving the lives of homeless and other disadvantaged people if their misfortunes can somehow be blamed on an inferior genetic endowment. Those in the forefront of genetics have a responsibility to acknowledge that human beings are more than the sum of their DNA sequences and an ethical obligation to ensure that the applications of new knowledge are beneficial to all.

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